

Variant: NM_001754.5(RUNX1):c.147A>G (p.Pro49=)

Version: 1.0

CA512318963 [↗](#)

2103402 (ClinVar) [↗](#)

Gene: RUNX1 ([HGNC:861](#))

Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome ([MONDO:0011071](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: d10cbbb7-ed35-481d-9cdb-621e77a37fc6

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HGVS expressions

NM_001754.5(RUNX1):c.147A>G (p.Pro49=)

NM_001754.5:c.147A>G

NC_000021.9:g.34887047T>C

CM000683.2:g.34887047T>C

NC_000021.8:g.36259344T>C

CM000683.1:g.36259344T>C

NC_000021.7:g.35181214T>C

NG_011402.2:g.1102665A>G

ENST00000675419.1:c.147A>G

ENST00000300305.7:c.147A>G

ENST00000344691.8:c.66A>G

ENST00000358356.9:c.66A>G

ENST00000399237.6:c.111A>G

ENST00000399240.5:c.66A>G

ENST00000437180.5:c.147A>G

ENST00000455571.5:c.108A>G

ENST00000475045.6:c.147A>G

ENST00000482318.5:c.59-6334A>G

NM_001001890.2:c.66A>G

NM_001122607.1:c.66A>G

NM_001754.4:c.147A>G

NM_001001890.3:c.66A>G

NM_001122607.2:c.66A>G

Likely Benign

Met criteria codes **3**

BP4 BP7 PM2_Supporting

Not Met criteria codes **23**

PS1 PS2 PS3 PS4 BP3 BP1
BP2 BP5 PP1 PP2 PP3 PP4
BA1 PM6 PM1 PM3 PM5
PM4 BS2 BS1 BS4 BS3
PVS1

Evidence Links **0**

Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Myeloid Malignancy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2

[↗](#) PDF

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.147A>G (p.Pro49=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.04) (BP4). This variant has a SpliceAI score ≤ 0.20 (0.04) and evolutionary conservation algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (-0.32) (BP7). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). In summary, this variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BP4, BP7, PM2_supporting.

Met criteria codes

BP4			This synonymous variant has a SpliceAI score ≤ 0.20 (0.04) (BP4).
BP7			This variant has a SpliceAI score ≤ 0.20 (0.04) and evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (-0.32) (BP7).
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).

Not Met criteria codes

PS1			This variant is not a missense variant.
PS2			De novo data for this variant has not been reported in literature.
PS3			In vitro or in vivo functional data has not been reported for this variant in the literature.
PS4			Proband data for this variant has not been reported in literature.
BP3			This rule is not applicable for MM-VCEP.
BP1			This rule is not applicable for MM-VCEP.
BP2			This variant has not been observed in trans with a pathogenic variant for a fully penetrant dominant gene/disorder or observed in cis with a pathogenic variant in any inheritance pattern.
BP5			This rule is not applicable for MM-VCEP.
PP1			Segregation data for this variant has not been reported in literature.

PP2		✖	This rule is not applicable for MM-VCEP.
PP3		✖	This synonymous or intronic variant does not have a REVEL score ≥ 0.88 or a SpliceAI score ≥ 0.38 .
PP4		✖	This rule is not applicable for MM-VCEP.
BA1		✖	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
PM6		✖	De novo data for this variant has not been reported in literature.
PM1		✖	This variant is not a missense variant.
PM3		✖	This rule is not applicable for MM-VCEP.
PM5		✖	This variant is not a missense variant.
PM4		✖	This variant is not an in-frame deletion/insertion.
BS2		✖	This rule is not applicable for MM-VCEP.
BS1		✖	This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS4		✖	Segregation data for this variant has not been reported in literature.
BS3		✖	In vitro or in vivo functional data has not been reported for this variant in the literature.
PVS1		✖	This variant is not a null variant.

Curation History 		
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